IN THE CLAIMS:

1-39. (cancelled)

40. **(allowed)** A method of detecting the presence in a human patient of an altered Survival Motor Neuron (SMN) gene associated with Spinal Muscular Atrophy, comprising:

analyzing exon 7 or exon 8 of a gene identified as T-BCD541 (SEQ ID NO:22) in a biological sample derived from the patient, and

comparing said exon 7 to the corresponding exon from nucleotide position 340 to nucleotide position 401 of SEQ ID NO:12, or exon 8 to the corresponding exon from nucleotide position 846 to nucleotide position 1408 of SEQ ID NO:12, which is present in a normal tissue;

wherein an alteration of either exon 7 or exon 8 in said patient sample with reference to said normal tissue is indicative of the presence of an altered Survival Motor Neuron (SMN) gene associated with Spinal Muscular Atrophy in said patient.

41. **(allowed)** The method of claim 40, wherein said analyzing comprises

determining whether T-BCD541 exon 7 is present or absent in the patient sample.

42. **(allowed)** The method of claim 40, wherein said analyzing comprises

determining whether T-BCD541 exon 8 is present or absent in the patient sample.

43. **(allowed)** The method of claim 40, wherein all or part of the T-BCD541 gene is subjected to PCR amplification prior to analyzing the gene for alterations in exon 7 or 8.

44. **(allowed)** The method of claim 43, wherein said analyzing comprises

amplifying a nucleotide fragment from said patient sample comprising exon 7 of the T-BCD541 gene,

amplifying a nucleotide fragment from said patient sample comprising exon 8 of the T-BCD541 gene, and

determining whether said exon 7 and said exon 8 are present or absent in said amplified fragments.

45. **(allowed)** The method of claim 44, wherein said determining includes

subjecting said exon 7 comprising nucleotide fragment to restriction enzyme digestion,

subjecting said exon 8 comprising nucleotide fragment to restriction enzyme digestion, and

analyzing enzymatic digestion products produced by said enzymatic digestions by comparing the enzymatic digestion products

from the biological sample to enzymatic digestion products of exon 7 or exon 8 of the survival motor neuron gene from normal tissue,

wherein an alteration of either exon 7 or exon 8 with reference to normal tissue is evidenced by an altered restriction enzymatic digestion pattern in one or both of said exons.

46. (presently amended) The method of claim 43, wherein said amplifying is carried out using a polymerase chain reaction using ~~a primer selected from the group consisting of~~ **the primer pairs consisting of** SEQ ID NO:5~~,~~ **and** SEQ ID NO:6, **or** SEQ ID NO:7 and SEQ ID NO:8**, respectively**.

47. **(allowed)** The method of claim 40, wherein said analyzing comprises subjecting said patient T-BCD541 gene to restriction cleavage with *Bsr*I, *Hind*III, *Sac*I or *Kpn*I.

48. **(allowed)** The method of claim 40, wherein said analyzing comprises subjecting said patient T-BCD541 gene present in said biological sample to single strand conformation polymorphism analysis, wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample to detect alterations in the patient gene.

49. **(allowed)** The method of claim 40, wherein said biological sample is selected from the group consisting of blood, cerebral fluid, peripheral blood leukocytes, a lymphoblastoid cell line and muscle tissue.

50. **(allowed)** A method of confirming a clinical diagnosis of Arthrogryposis Multiplex Congenita in a patient, comprising

analyzing exon 7 or exon 8 of a gene identified as T-BCD541 (SEQ ID NO:22) in a biological sample derived from the patient, and

comparing said exon 7 to the corresponding exon from nucleotide position 340 to nucleotide position 401 of SEQ ID NO:13, or exon 8 to the corresponding exon from nucleotide position 846 to nucleotide position 1408 of SEQ ID NO:13, which is present in a normal tissue;

wherein an alteration of either exon 7 or exon 8 in said patient sample with reference to said normal tissue is indicative of the presence of an altered Survival Motor Neuron (SMN) gene associated with Arthrogryposis Multiplex Congenita in said patient.

51. **(allowed)** The method of claim 50, wherein said analyzing comprises

amplifying a nucleotide fragment from said patient sample comprising exon 7 of the T-BCD541 gene,

amplifying a nucleotide fragment of said patient sample comprising exon 8 of the T-BCD541 gene, and

determining whether said exon 7 and said exon 8 are present or absent in said amplified nucleotide fragments.

52. **(allowed)** The method of claim 51, wherein said determining includes

subjecting said exon 7 comprising nucleotide fragment to restriction enzyme digestion,

subjecting said exon 8 comprising nucleotide fragment to restriction enzyme digestion, and

analyzing enzymatic digestion products produced by said enzymatic digestions by comparing the enzymatic digestion products from the biological sample to enzymatic digestion products of exon 7 or exon 8 of the survival motor neuron gene from normal tissue,

wherein an alteration of either exon 7 or exon 8 with reference to normal tissue is evidenced by an altered restriction enzymatic digestion pattern in one or both of said exons.

53. (cancelled)

54. **(allowed)** A method of identifying the presence or absence of a mutation in the Survival Motor Neuron (SMN) gene of SEQ ID NO:22 in a nucleic acid sample, comprising

(a) subjecting the nucleic acid in the sample to digestion by a restriction endonuclease, wherein restriction fragments

resulting from said digestion of a mutated SMN gene differ from those obtained from a T-BCD541 gene of SEQ ID NO: 12; and

(b) identifying the presence or absence of a mutation in the SMN gene in the subject.

55. **(allowed)** The method of claim 54, wherein the restriction endonuclease is *Bsr-1*.

56. (presently amended) The method of claim 54, **further comprising isolating the nucleic acid after step (a) and subjecting the nucleic acid** ~~wherein the nucleic acid is further subjected~~ to a polymerise chain reaction (PCR) following isolation.

57. (cancelled)

58. (presently amended) A method of identifying the presence of Spinal Muscular Atrophy (SMA) in a subject, said method comprising:

(a) identifying a mutation in a T-BCD541 gene **consisting of SEQ ID NO:22** ~~(SEQ ID NO:22)~~ in a DNA sample obtained from said subject;

wherein the presence of a mutation in the T-BCD541 gene is indicative of the presence of SMA in said subject.

59. (presently amended) The method of claim 58, wherein the mutation is a deletion in the T-BCD541 gene **consisting of SEQ ID NO:22** ~~(SEQ ID NO:22)~~.

60. (presently amended) The method of claim 59, wherein the deletion comprises a deletion of the entire T-BCD541 gene **consisting of SEQ ID NO:22** ~~(SEQ ID NO:22)~~.

61. (presently amended) **A method of identifying the presence of Spinal Muscular Atrophy (SMA) in a subject, said method comprising identifying a mutation in a T-BCD541 gene consisting of SEQ ID NO:22 in a DNA sample obtained from said subject, wherein the presence of a mutation that** ~~The method of claim 59, wherein the mutation~~ results in a truncation of the protein product encoded by SEQ ID NO:12 **is indicative of the presence of SMA in said subject**.

62. (presently amended) The method of claim 58, wherein the sequence of the isolated nucleic acid is determined by ~~direct~~ **nucleotide** sequencing.

63. (presently amended) The method of claim 58, **further comprising isolating the nucleic acid after step (a) and subjecting the nucleic acid** ~~wherein the nucleic acid is further subjected~~ to polymerise chain reaction (PCR) following isolation.

64-67. (cancelled)

68. (presently amended) A method for detecting the presence or absence of Spinal Muscular Atrophy in an individual, comprising the steps of:

(a) contacting a biological test sample obtained from the individual with a nucleic acid probe comprising ~~all or part of~~ SEQ ID NO: 12 or 13, ~~or a complement of SEQ ID NO: 12 or 13,~~ wherein the nucleic acid probe detects a truncation, deletion or mutation of SEQ ID NO: 12 or 13,

(b) maintaining the test sample and the nucleic acid probe under conditions suitable for hybridization;

(c) detecting hybridization between the test sample and probe; and

(d) comparing hybridization in the test sample to a control sample, wherein no detectable hybridization between the test sample and probe is indicative of the presence of Spinal Muscular Atrophy in the individual.

69. (presently amended) A method for detecting the presence or absence of Spinal Muscular Atrophy in an individual, comprising analyzing a DNA sample obtained from the individual, wherein the DNA sample comprises the Survival Motor Neuron gene and wherein the method comprises detecting the presence or absence of either exon 7 or exon 8, or both exon 7 and exon 8 of the gene, wherein exon 7 ~~comprises~~ **consists of** nucleotides 340 to 401 of SEQ ID NO: 13, and exon 8 ~~comprises~~ **consists of** nucleotides 846 to 1408 of SEQ NO: 13, wherein the absence of either or both exon 7 or 8 is indicative of the presence of Spinal Muscular Atrophy in the individual.

70. (cancelled)